# KRT3 gene

keratin 3

## Normal Function

The *KRT3* gene provides instructions for making a protein called keratin 3. Keratins are a group of tough, fibrous proteins that form the structural framework of epithelial cells, which are cells that line the surfaces and cavities of the body. Keratin 3 is produced in a tissue on the surface of the eye called the corneal epithelium. This tissue forms the outermost layer of the cornea, which is the clear front covering of the eye. The corneal epithelium acts as a barrier to help prevent foreign materials, such as dust and bacteria, from entering the eye.

The keratin 3 protein partners with another keratin protein, keratin 12, to form molecules known as intermediate filaments. These filaments assemble into strong networks that provide strength and resilience to the corneal epithelium.

## Health Conditions Related to Genetic Changes

### Meesmann corneal dystrophy

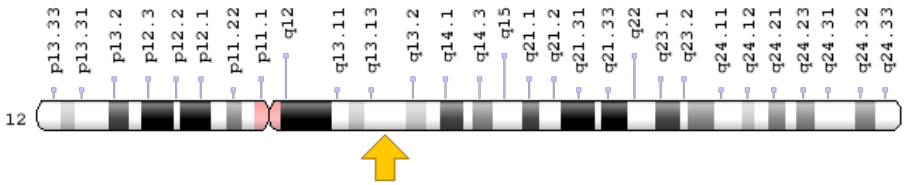
At least three mutations in the *KRT3* gene have been found to cause Meesmann corneal dystrophy, an eye disease characterized by the formation of tiny cysts in the corneal epithelium.

All of the identified *KRT3* gene mutations associated with Meesmann corneal dystrophy change single protein building blocks (amino acids) in the keratin 3 protein. These changes occur in a region of the protein that is critical for the formation and stability of intermediate filaments. The altered keratin 3 protein interferes with the assembly of intermediate filaments, weakening the structural framework of the corneal epithelium. As a result, this outer layer of the cornea is abnormally fragile and develops the cysts that characterize Meesmann corneal dystrophy. The cysts likely contain clumps of abnormal keratin proteins and other cellular debris. When the cysts break open (rupture), they cause eye irritation, increased sensitivity to light (photophobia), and related symptoms.

## Chromosomal Location

Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Molecular Location: base pairs 52,789,685 to 52,796,108 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 65 kDa cytokeratin
- CK-3
- CK3
- cytokeratin 3
- cytokeratin-3
- K2C3_HUMAN
- K3
- keratin 3, type II
- keratin, type II cytoskeletal 3
- type-II keratin Kb3

## Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Intermediate Filaments https://www.ncbi.nlm.nih.gov/books/NBK21560/
- National Eye Institute: Facts About the Cornea and Corneal Disease https://nei.nih.gov/health/cornealdisease/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28KRT3%5BTIAB%5D%29+OR+%28keratin+3%5BTIAB%5D%29%29+OR+%28%28CK-3%5BTIAB%5D%29+OR+%28cytokeratin+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- KERATIN 3, TYPE II
  http://omim.org/entry/148043

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KRT3%5Bgene%5D
- HGNC Gene Family: Keratins, type II
  http://www.genenames.org/cgi-bin/genefamilies/set/609
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6440
- Human Intermediate Filament Database
  http://www.interfil.org/details.php?id=NM_057088
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3850
- UniProt
  http://www.uniprot.org/uniprot/P12035

## Sources for This Summary


- Chen YT, Tseng SH, Chao SC. Novel mutations in the helix termination motif of keratin 3 and keratin 12 in 2 Taiwanese families with Meesmann corneal dystrophy. Cornea. 2005 Nov;24(8):928-32.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/16227835
- Irvine AD, Corden LD, Swensson O, Swensson B, Moore JE, Frazer DG, Smith FJ, Knowlton RG, Christophers E, Rochels R, Uitto J, McLean WH. Mutations in cornea-specific keratin K3 or K12 genes cause Meesmann's corneal dystrophy. Nat Genet. 1997 Jun;16(2):184-7.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/9171831
- Szaflik JP, Oldak M, Maksym RB, Kaminska A, Pollak A, Udziela M, Ploski R, Szaflik J. Genetics of Meesmann corneal dystrophy: a novel mutation in the keratin 3 gene in an asymptomatic family suggests genotype-phenotype correlation. Mol Vis. 2008 Sep 15;14:1713-8.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/18806880
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2538492/

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